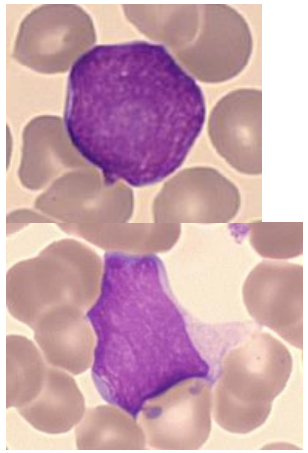
Cryptique ou non?

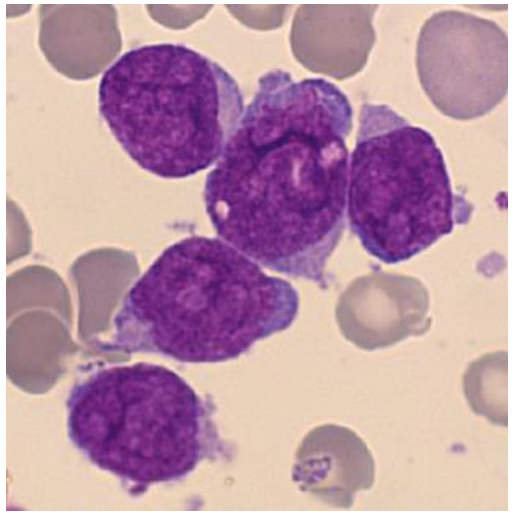
Lucie Coster

CHU de Toulouse

Homme de 20 ans : dyspnée, toux sèche, douleur thoracique, masse médiastinale

Hémoglobine	15.6	g/dL
VGM	83.8	fL
TGMHb	29.4	pg
Hématocrite	44.5	%
Hématies	5.31	T/L
CGMHb	35.1	g/dL
Indice de distribution	13.4	%
Technique: Automate Sysmex XN		
Plaquettes	178	G/L
VMP	10.3	fL
Technique: Automate Sysmex XN		
Leucocytes	9.98	G/L
Polynucléaires neutrophiles	57.3	%
	5.7	G/L
Polynucléaires éosinophiles	2.2	%
	0.2	G/L
Polynucléaires basophiles	0.0	%
	0.0	G/L
Lymphocytes	14.8	%
	1.5	G/L
Monocytes	3.7	%
	0.4	G/L
Blastes	22	%
	2.2	G/L

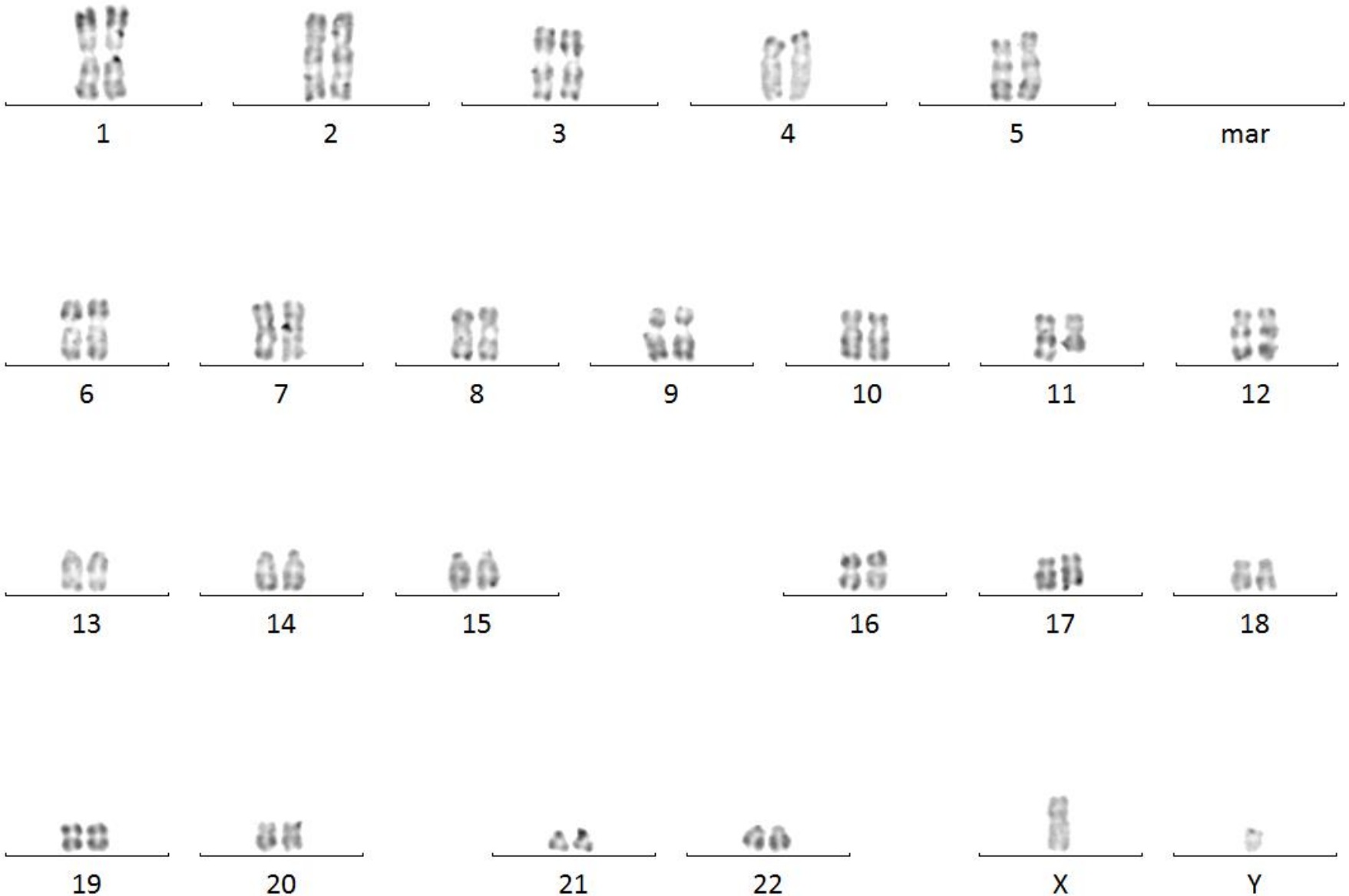




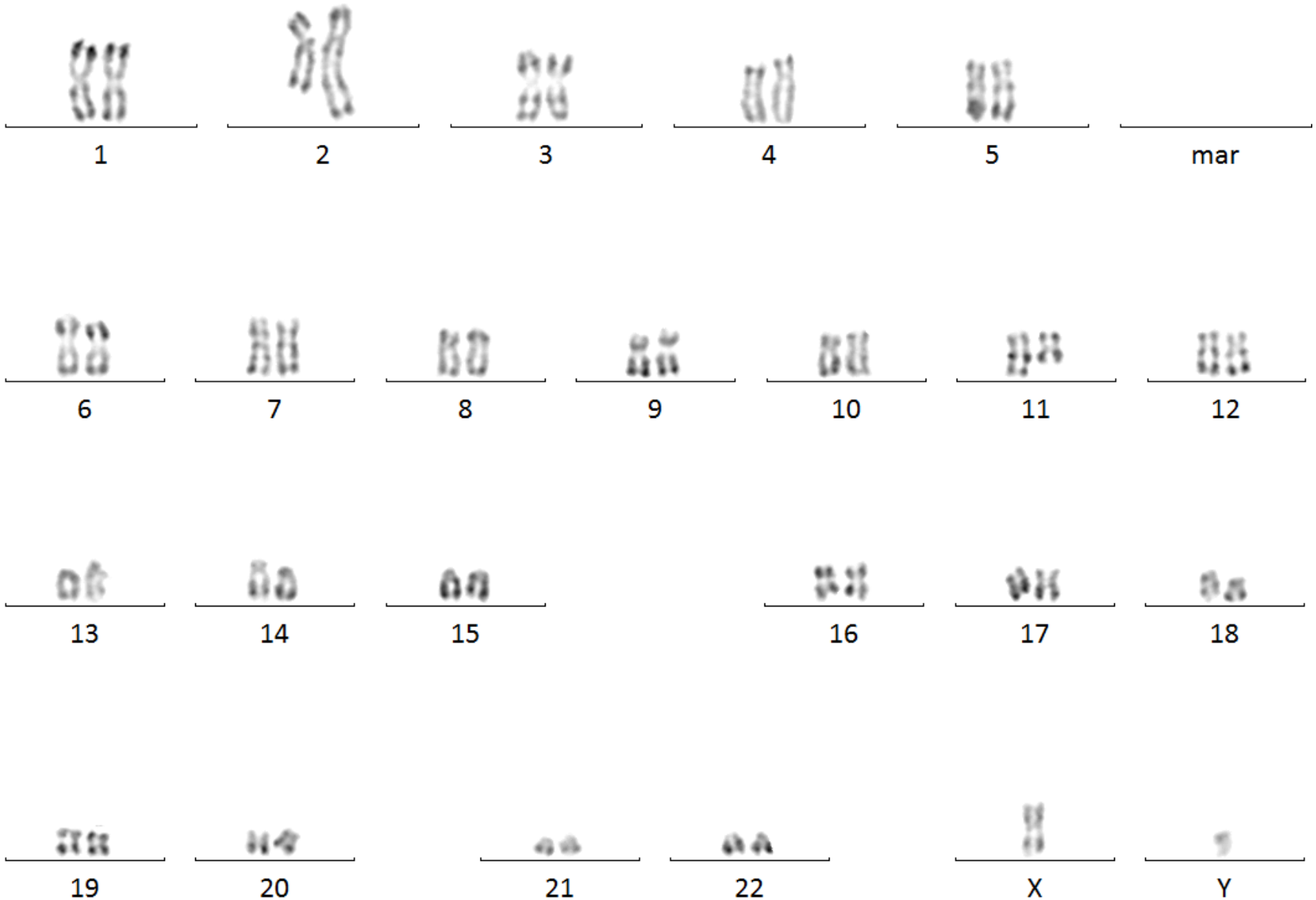
Nature du prélèvement : Sang EDTA

Marqueurs de membranes											
Lignée B (%)			Lignée T (%)			Myéloide (%)			Autres (%)		
CD19	:	0	CD1a	:	10	CD13	:	51	CD34	:	95
CD20	:	#nf	CD2	:	3	CD33	:	19	HLA-DR	:	#nf
CD22	:	#nf	CD3	:	0.4	CD117	:	53	CD56	:	2
CD10	:	32	CD4	:	18	CD34+13+	:	48		:	
CD19+CD10+	:	#nf	CD8	:	2	CD34+117+	:	46		:	
			CD5	:	85		:				
			CD7	:	99		:				
			CD3+TCRαβ+	:	3		:				
			CD3+TCRγδ+	:	2		:				
Marqueurs cytoplasmiques											
Lignée B (%)			Lignée T (%)			Myéloide (%)			Autres (%)		
IgMc	:	0	CD3c	:	98	MPOc	:	0		:	
CD22c	:	#er		:			:			:	
CD79a	:	0		:			:			:	

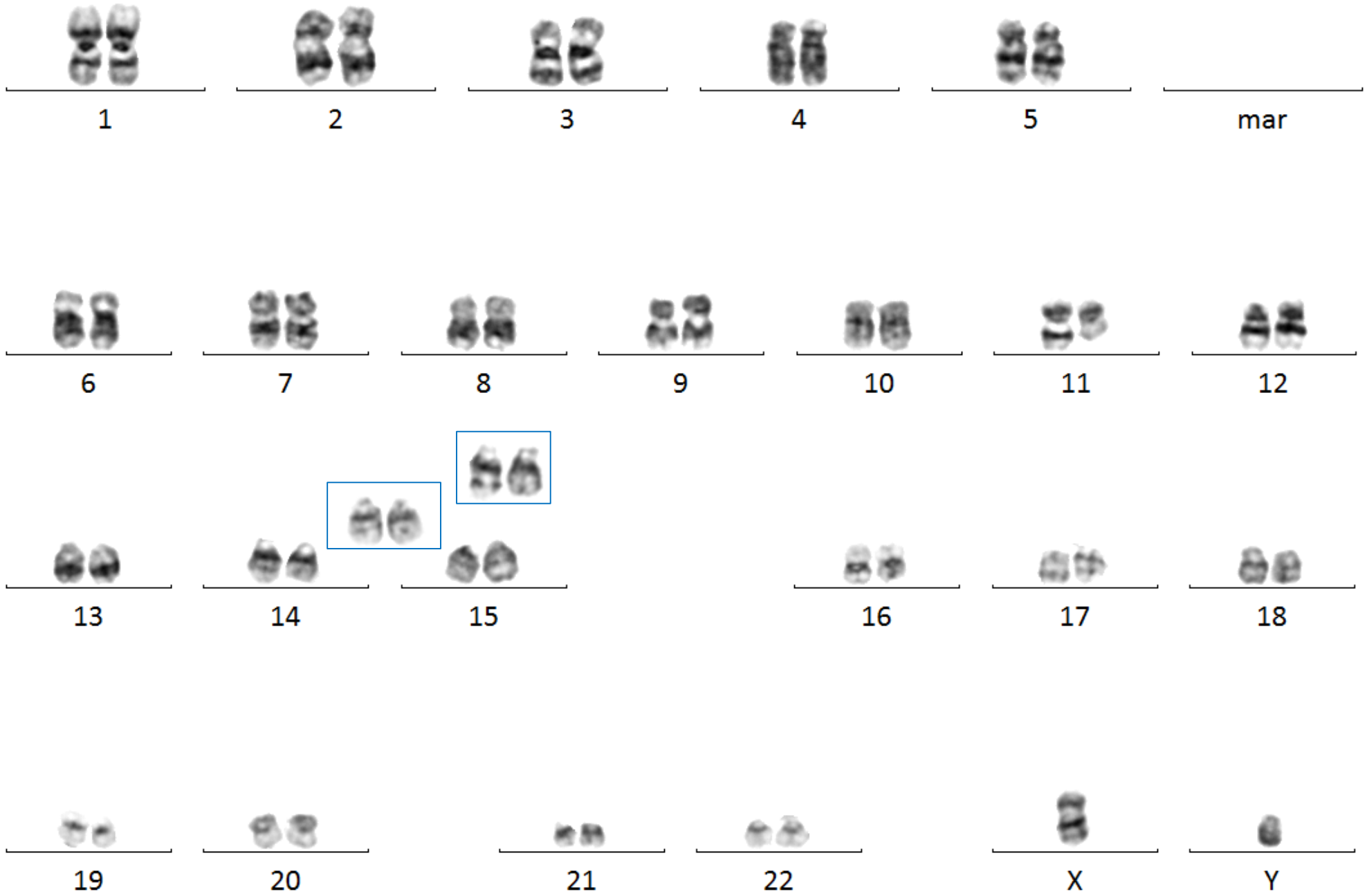
CONCLUSION: Prolifération lymphoblastique T de type LAL near-ETP (OMS 2016).



11/20 mitoses



3/20 mitoses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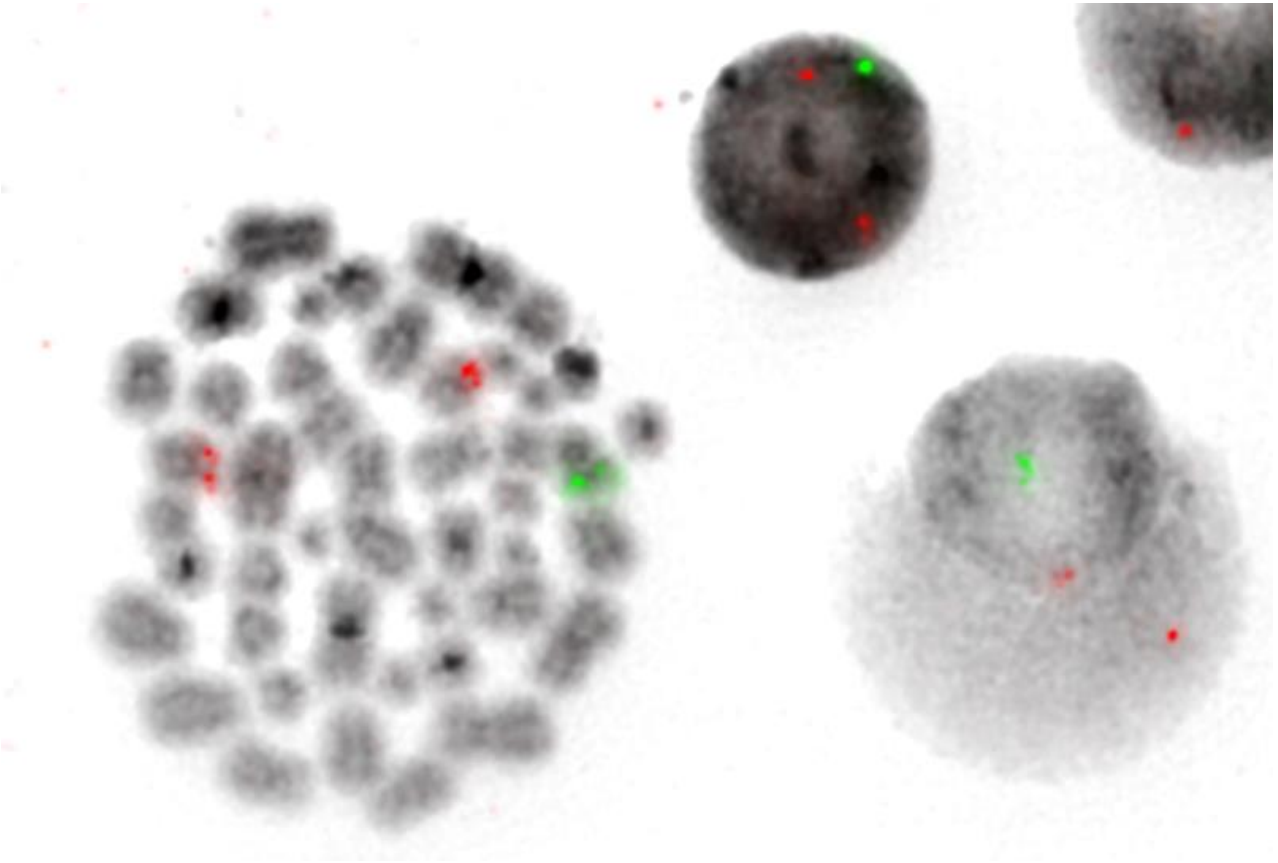
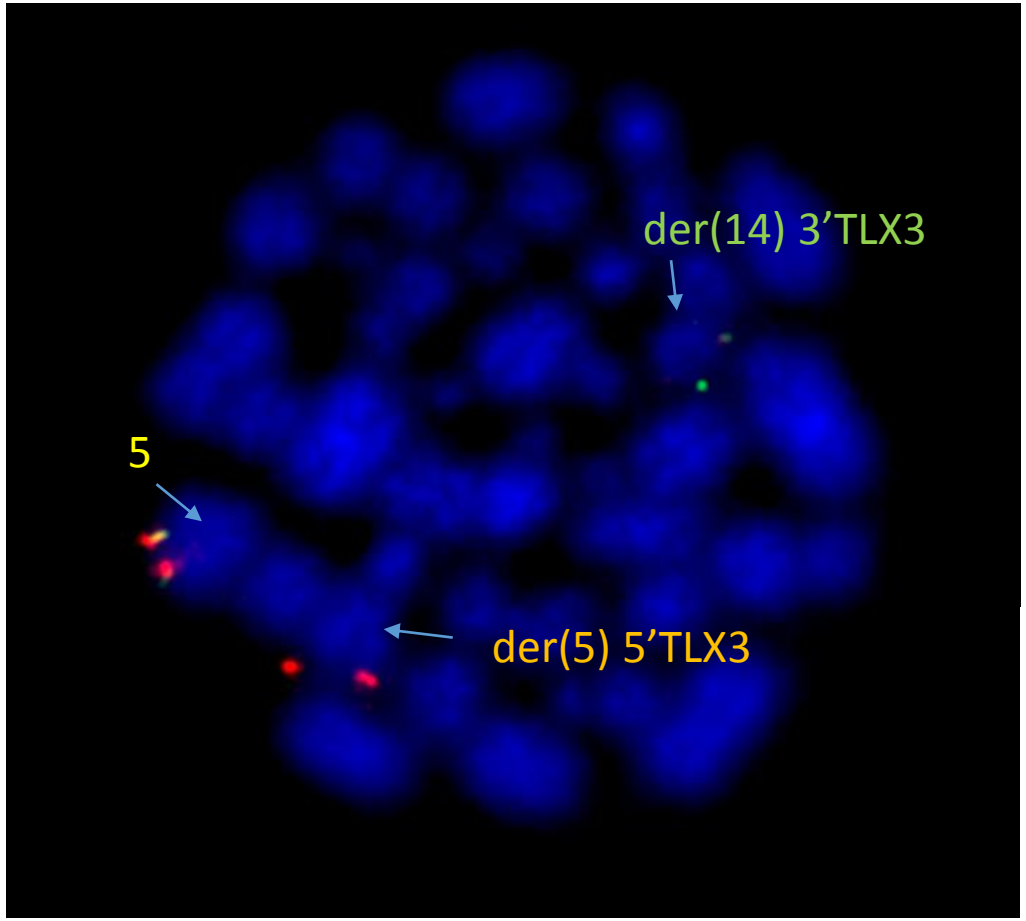
FISH PICALM-MLLT10 maison

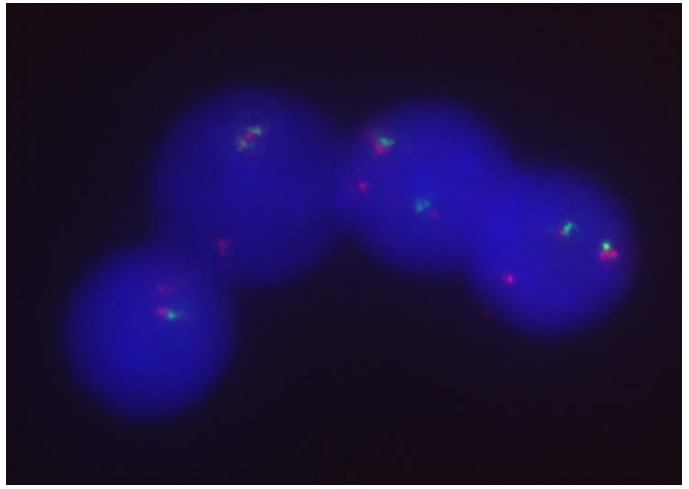
Absence de réarrangement PICALM-MLLT10.
Perte d'une copie du gène PICALM dans 20/20 mitoses et 90% des noyaux



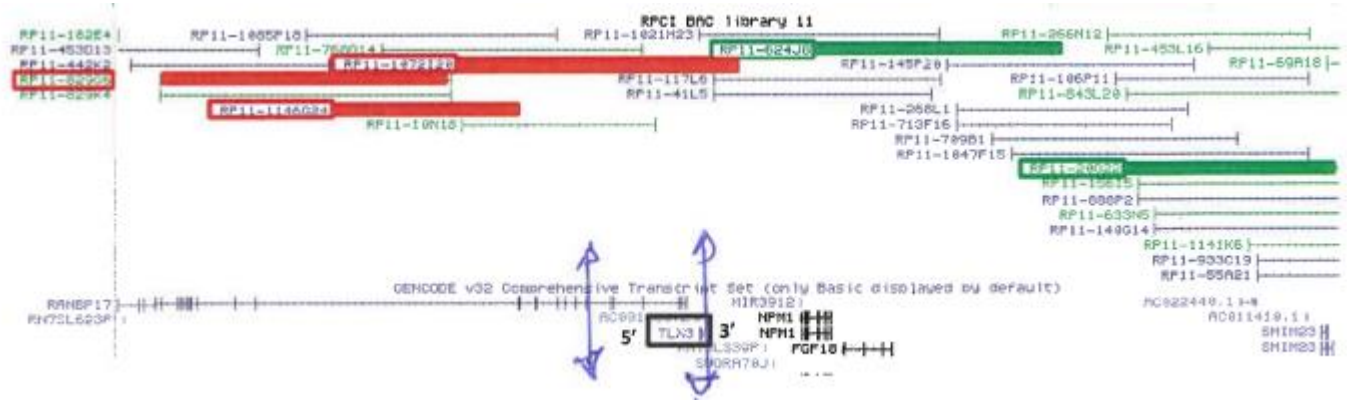
FISH BCR-ABL1, TLX1 et KMT2A normales

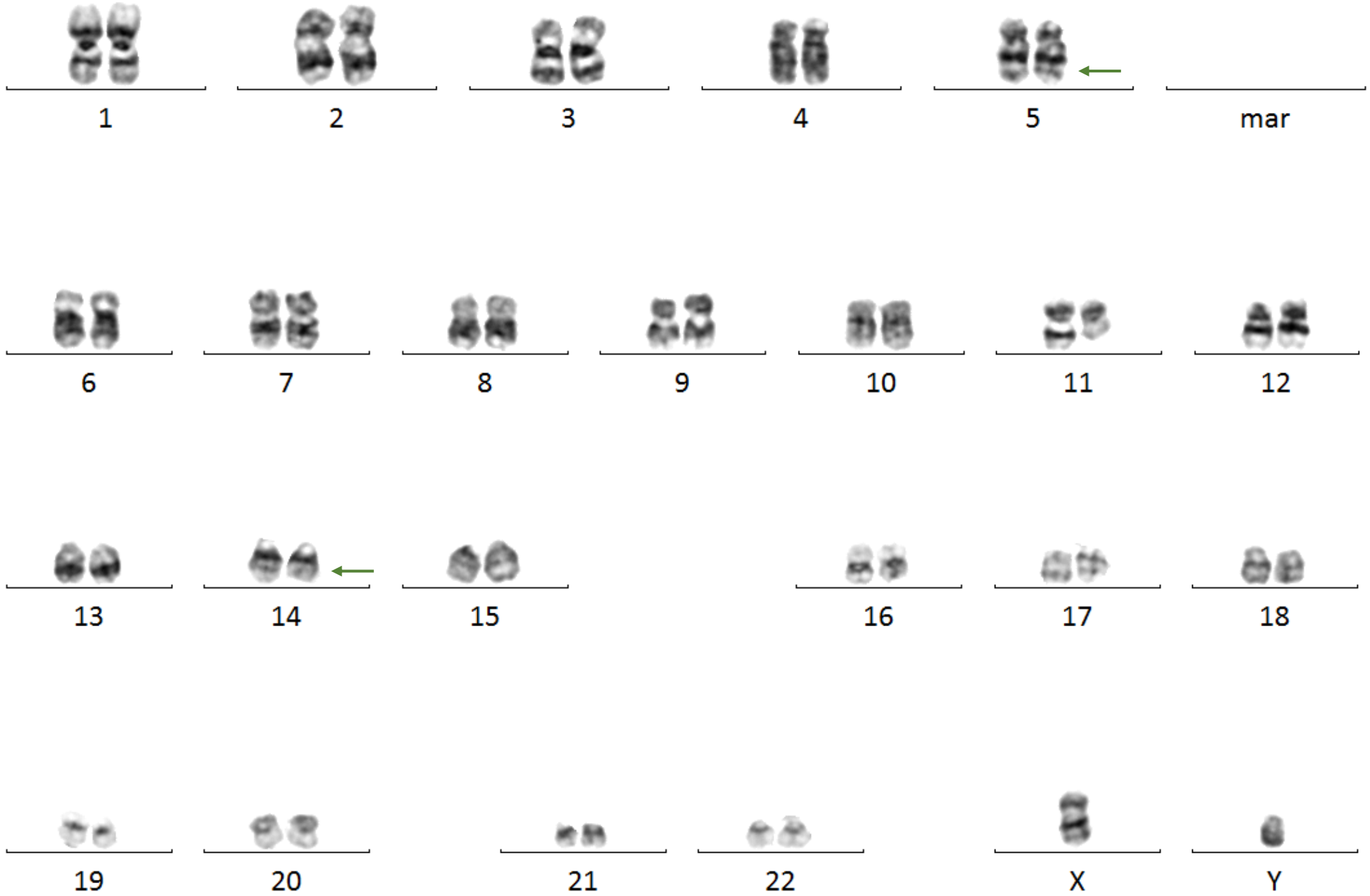
FISH TLX3 (5q35) double couleur maison





Centromère ⟵ ⟶ Télomère

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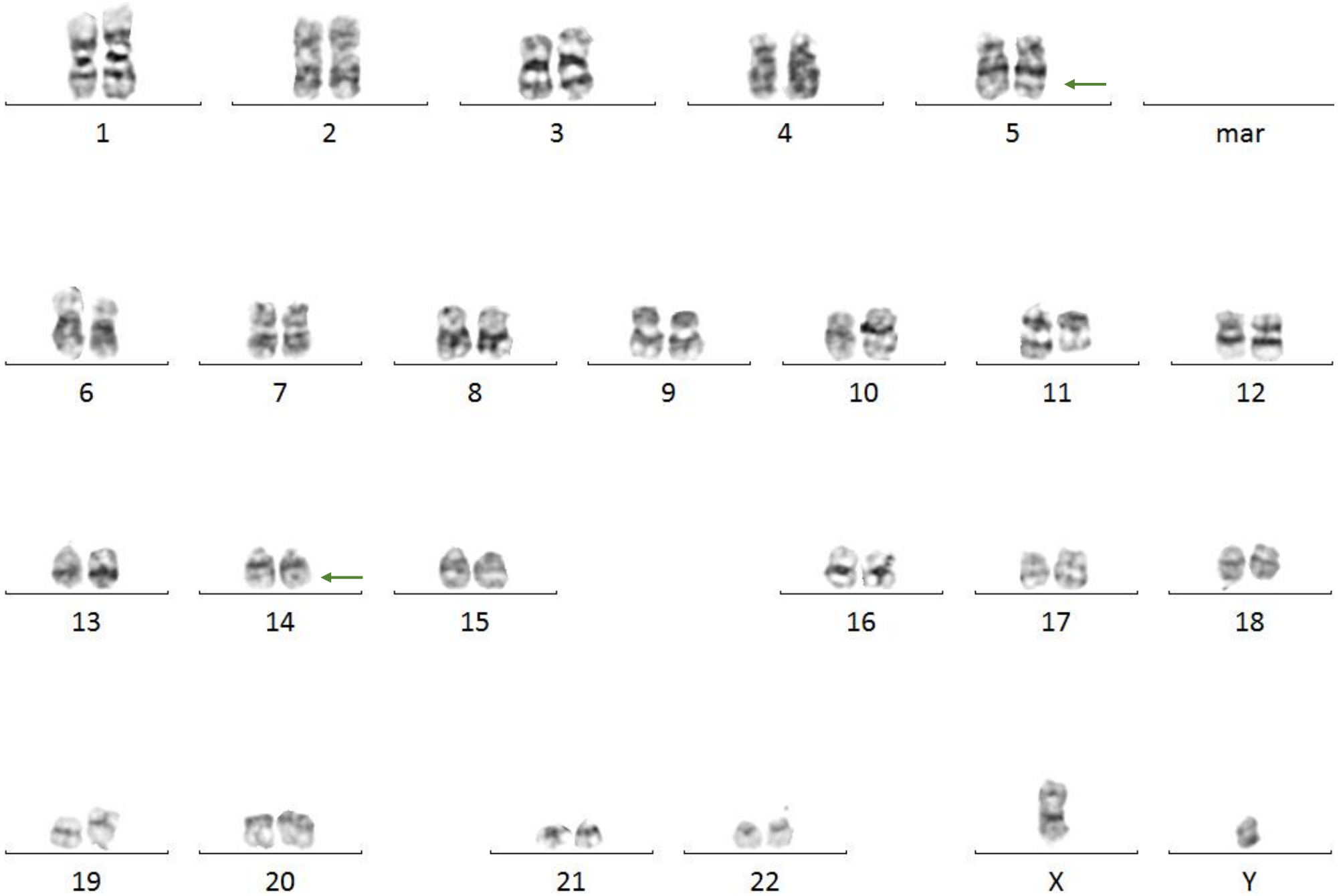
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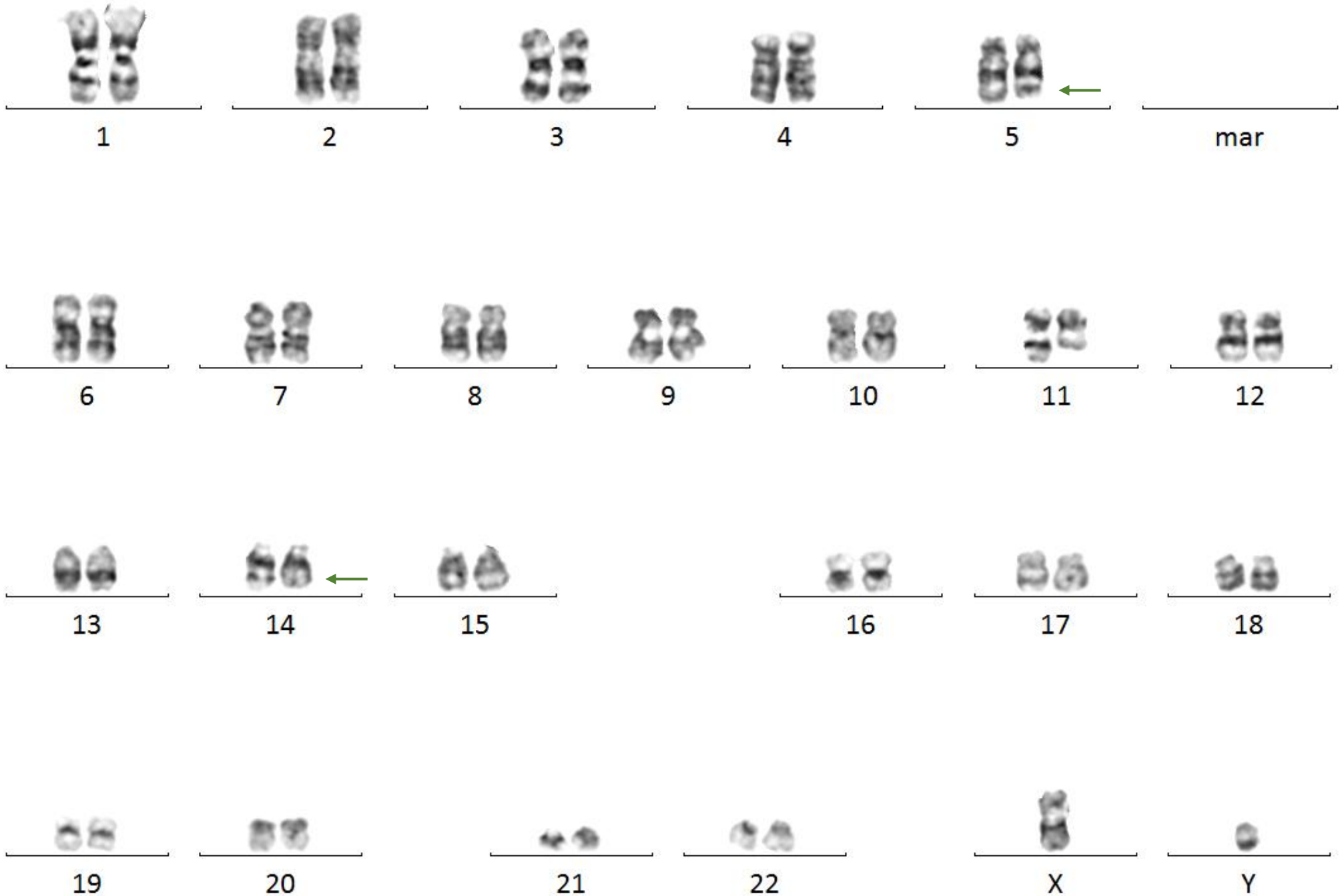
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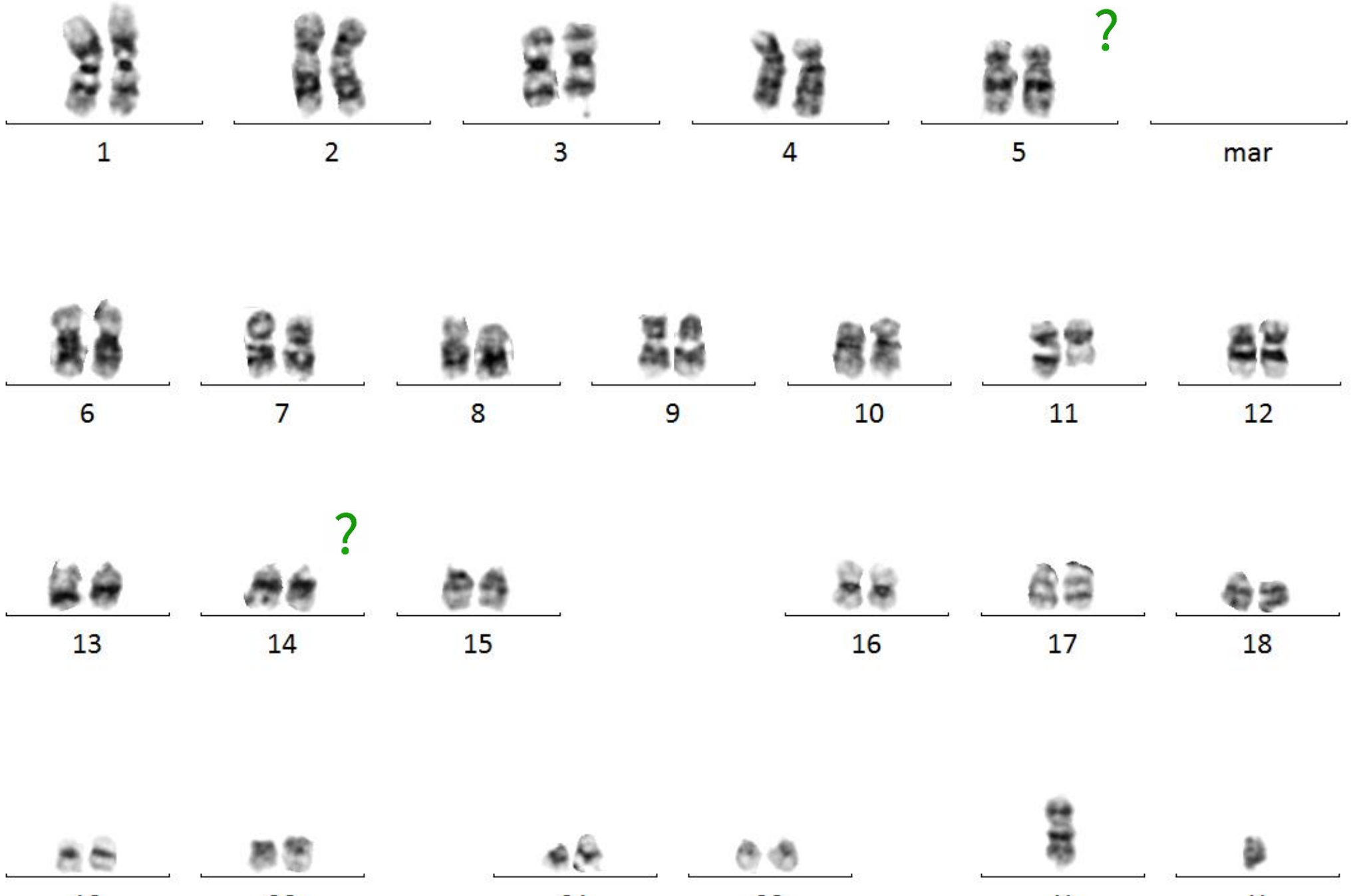
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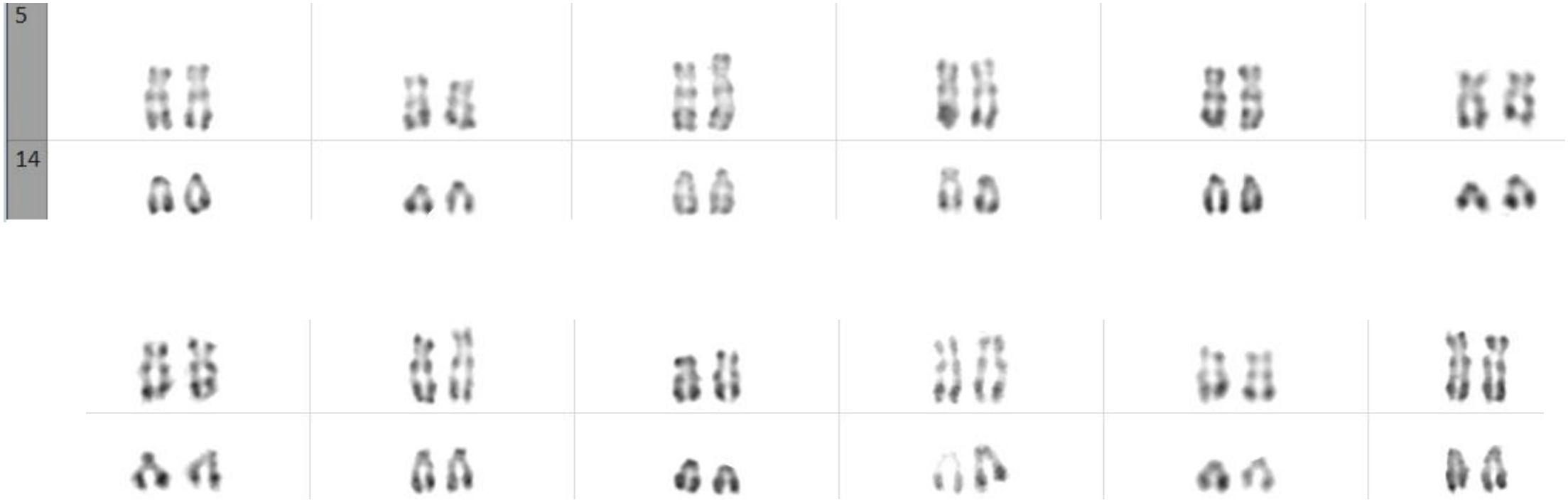
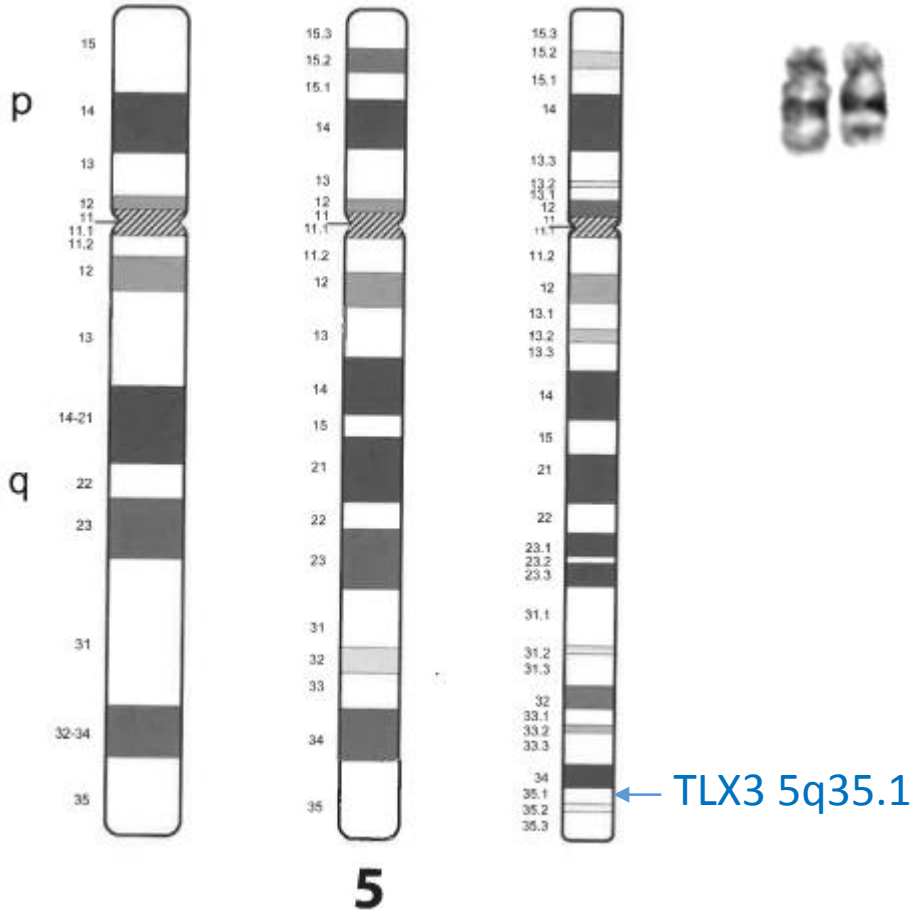
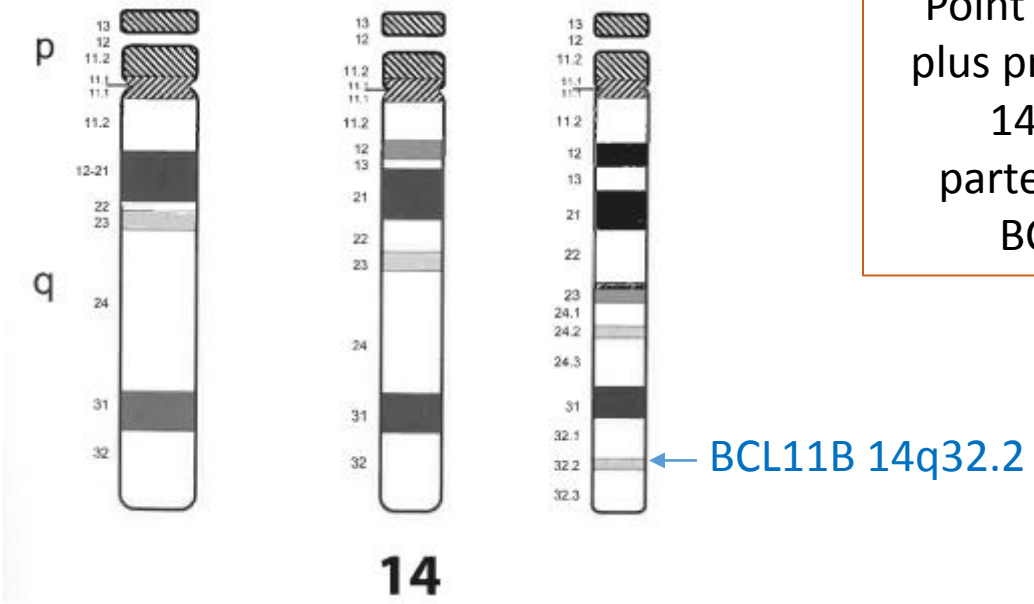
Retour sur les bandes RHG



(5 et 14 des mitoses anormales)

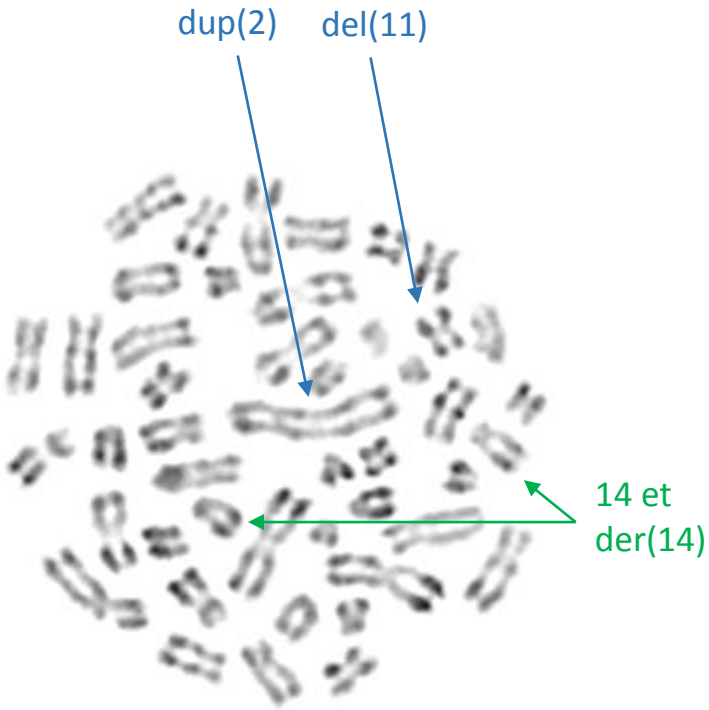
t(5;14) TLX3/BCL11B : points de cassure

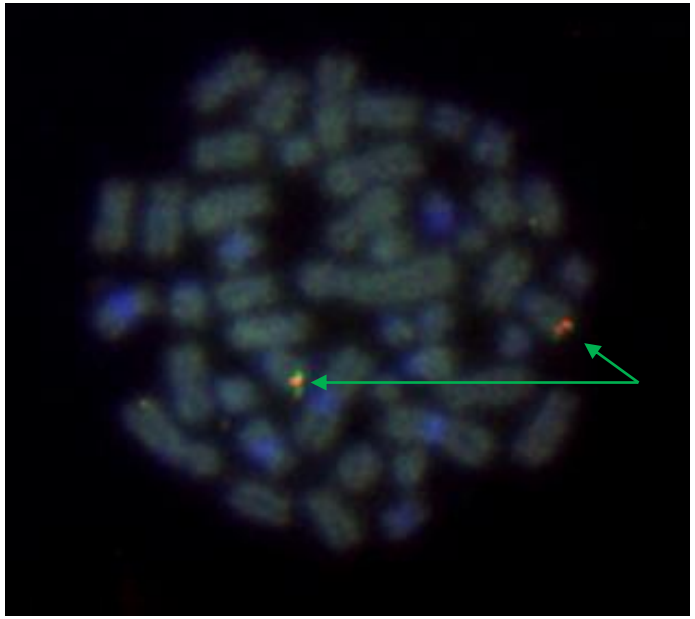




Point de cassure plus proximal que 14q32.2 / partenaire non BCL11B?

FISH TCL1A (14q32.13) double couleur maison sur lame RHG



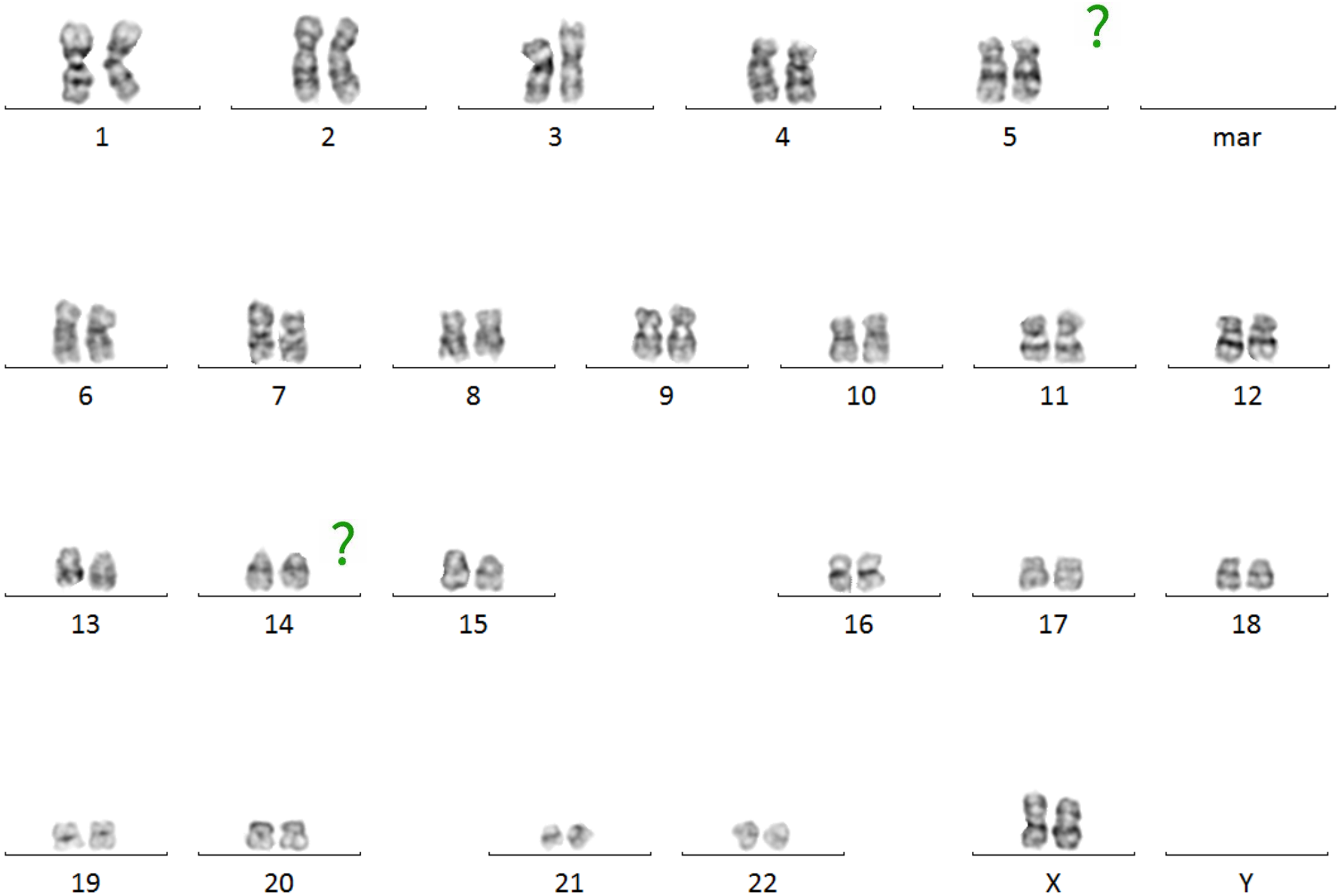


Point de cassure plus télomérique que TCL1A :
Compatible avec un réarrangement de BCL11B (14q32.2)

Patient 2

46,XX[20].ish t(5;14)(5'TLX3+;3'TLX3+)[18],9q34(ABLx2),22q11(BCRx2)[2], 10q24(TLX1x2)[2].nuc ish (TLX3x2)(5'TLX3 sep 3'TLX3x1) [179/200],(ABL,BCR)x2<200>,(TLX1x2)[200]

(5 et 14 de toutes les mitoses)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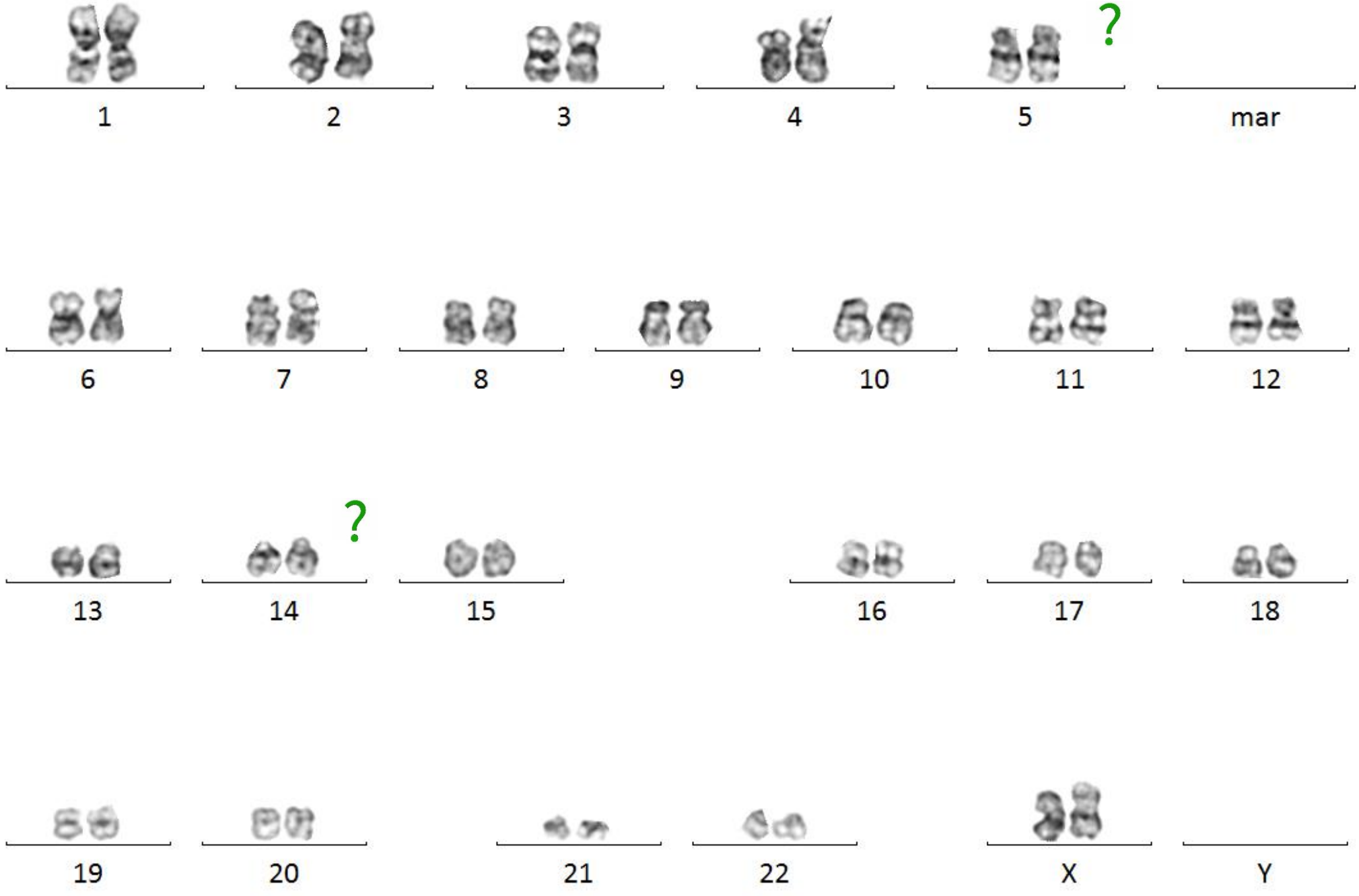
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Patient 3

46,X,-Y,add(4)(q2?5),der(6)t(?4,6)(?q25;p21),+mar[13]/ 47,sl,+19[3]/46,XY[4].ish t(5;14)(q35;q32)(5'TLX3-;3'TLX3+)[5],9q34(ABLx2), 22q11(BCRx2)[3],mar

(wcp8-)[9].nuc ish(ABL,BCR)x2[200]

(5 et 14 des mitoses anormales)

Au final…

- Cryptique ou non?
  - t(5;14) visible en bandes G, à la faveur d'une bonne résolution?
  - Ou… délétion 14q associée à la translocation t(5;14) chez le patient 1?
- Intérêt des bandes G pour la détection d'anomalies chromosomiques

Merci pour votre attention